Disorders Detectable by Tandem Mass Spectrometry (MS/MS) Using Newborn Screening Dried Blood Spots*

Amino Acid Disorders

- maple syrup urine disease (MSUD)
- phenylketonuria (PKU)
- citrullinemia/argininosuccinic acid synthetase deficiency (ASAS deficiency)
- homocystinuria/cystathionine beta-synthase deficiency (CBS deficiency)
- argininosuccinyl-CoA lyase deficiency (ASAL deficiency)
- argininemia/arginase deficiency
- tyrosinemia

Organic Acid Disorders

- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA lyase deficiency)
- glutaric acidemia type-1 (GA-1)
- isobutyryl-CoA dehydrogenase deficiency
- isovaleric acidemia (IVA)
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC deficiency)
- propionic acidemia (PA)
- methylmalonic acidemia (MMA)
- beta-ketothiolase deficiency (BKD)

Fatty Acid Oxidation Disorders

- carnitine transporter deficiency
- carnitine-acylcarnitine translocase deficiency (CAT deficiency)
- multiple acyl-CoA dehydrogenase deficiency (MAD deficiency)/glutaric acidemia type-2 (GA-2)
- 3-hydroxy long chain acyl-CoA dehydrogenase deficiency (LCHAD deficiency)/ trifunctional protein deficiency (TFP deficiency)
- medium chain acyl-CoA dehydrogenase deficiency (MCAD deficiency)
- carnitine palmitoyl transferase deficiency-type 1 (CPT-1 deficiency)
- carnitine palmitoyl transferase deficiency-type 2 (CPT-2 deficiency)
- short chain acyl-CoA dehydrogenase deficiency (SCAD deficiency)
- very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency)

****Note: This 1/15/02 revised list replaces the earlier 11/13/01 version. As with the earlier version, some of these listed disorders may not be included in the California Department of Health Services, Tandem Mass Spectrometry (MS/MS) Research Project but should be reported by any health care provider making a diagnosis on or after 1/7/02.***